

# 1994 ASHG PRESIDENTIAL ADDRESS

## Who Are We? Where Are We Going? Anticipating the 21st Century

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There is, I believe, general agreement that interest and activity in human genetics has today reached a peak never before attained. The periodical literature of the last ten years and the reports of the increasingly frequent symposia and conferences devoted to genetic problems in man provide convincing evidence of this. It is also clear that interest in these problems is likely to increase greatly in the next years so that what we may be witnessing now is only the beginning of a kind of renaissance in which genetics in general stands a chance of being greatly enriched by research on man.

These words were originally spoken by L. C. Dunn, in his presidential address entitled "Cross Currents in the History of Human Genetics," to the 1961 annual meeting of The American Society of Human Genetics (the "Society") (Dunn 1961). These sentiments are as true and relevant today as they were 33 years ago. Incidentally, that was the first meeting of the Society that I attended and the one at which I made my first scientific presentation. It was the first paper of the first morning session, and the audience was quite sparse, since most of the participants were mesmerized by the television coverage of Alan Sheppard, who was about to be launched in the first suborbital flight of the U.S. space program. That meeting was held in conjunction with the Society for Pediatric Research, and at the plenary session there were perhaps 250–300 people representing both organizations, a far cry from the present assembly in this auditorium, which holds 5,200. There are 4,279 registrants at this meeting, a better than 20-fold increase in Society attendance in the intervening 30 years.

In past decade or 15 years, tremendous changes have occurred in the scope and breadth of the Society's activities. Indeed, the entire genetics community has undergone significant expansion, giving rise to numerous professional organizations, governmental agencies, and lay consumer/advocacy groups, all of which have slightly different interests and missions within the discipline of human genetics.

This rapid expansion of activities and services has been in response principally to internal forces:

- *research developments* through our own efforts, leading to enhanced services and creating a need for more practitioners and trainees;
- broadening of the stream of genetic information to the lay public, leading to *increased consumer awareness*;
- an appreciation of the *lack of personnel* trained to transmit this increasing genetic information to both professionals and consumers, leading to the emergence of the genetic counselor or associate as a new genetic-health-care professional;
- the recognition of a *growing shortage of resources*, a diminished or redirected trainee pool, and an increased demand for educational efforts to a much broader audience.

### What Has Happened to the Society Itself during the Past Decade?

The membership has doubled to its present level of 5,800, an 8.2% annual growth rate, with no apparent indication of deceleration (table 1). This rate projects attaining a membership of 6,000 by the end of 1994. Interestingly, a significant proportion of this growth is due to international membership, with 15% of its membership being derived from countries other than the United States, Canada, and Mexico. The greatest overseas membership increase in the past 10 years has come from Europe and Asia (table 2).

As twice before, the Society has recently undertaken a survey to describe the demographic makeup, specialty distribution, training programs, and salary scales of its members. This time, however, the Society component is part of a much broader effort under the sponsorship of the Council of Regional Genetics Networks (CORN) and the Maternal and Child Health Branch of the Department of Health and Human Services. Never before has there been an attempt to develop as a broad database with responses being solicited from so many different groups. A number of different questionnaires were sent to Society members, chairs and chiefs of academic departments or divisions, genetics laboratory directors, and others who provide genetic services. I would like to take this opportunity to

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**Table 1****ASHG Membership Trends in the Past Decade**

Year	Total Members	% Increase
1985 .....	2,892	
1986 .....	3,163	9.3
1987 .....	3,434	8.6
1988 .....	3,769	9.8
1989 .....	4,140	6.2
1990 .....	4,399	2.8
1991 .....	4,520	7.3
1992 .....	4,961	9.8
1993 .....	5,321	7.3
Projected:		
1994 .....	6,000	12.8
1995 .....	6,300	

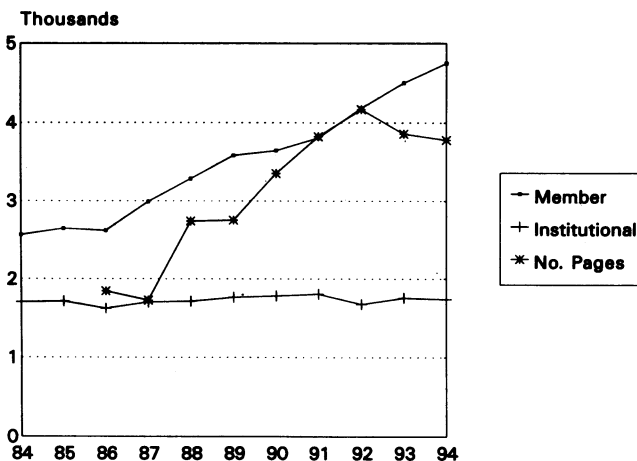
thank all of you who completed and returned these questionnaires, which demanded a significant investment of your time. I would like to give special thanks to Anne Smith, Secretary of the Society, and Jessica Davis, outgoing president of CORN, who initiated this project and have worked on it for some time.

Although data analysis is not yet complete, certain preliminary observations can be provided at this time, covering the Society component. Based on the responses received, it is clear that the Society is maturing. The age distribution indicates an increase of almost 2 years in the mean age of the membership in 1994 (43.2 years), compared to 41.7 in 1984 and 41.3 in 1989. This observation is supported by the professional ranks held by those members in academic institutions. There is almost equal distribution among the assistant (27%), associate (24%), and full professors (29%), demonstrating an increase in the tenured faculty among the membership.

Comparison of the primary areas of research involvement in the current survey with the previous ones indicates an increase in clinical activities (of both a basic and applied

**Table 2****Geographic Distribution of ASHG Members**

	1983		1994	
	No.	%	No.	%
North America .....	1,859	93	4,730	85.0
South/Central America .....	16	1	55	1.0
Europe .....	69	3	455	8.2
Africa .....	11	1	18	.3
Asia .....	20	1	160	2.9
Middle East .....	17	1	74	1.3
Pacific Rim .....	14	1	76	1.5
Total .....	2,006		5,568	



**Figure 1** Yearly subscription totals and pages devoted to scientific papers for *The American Journal of Human Genetics*.

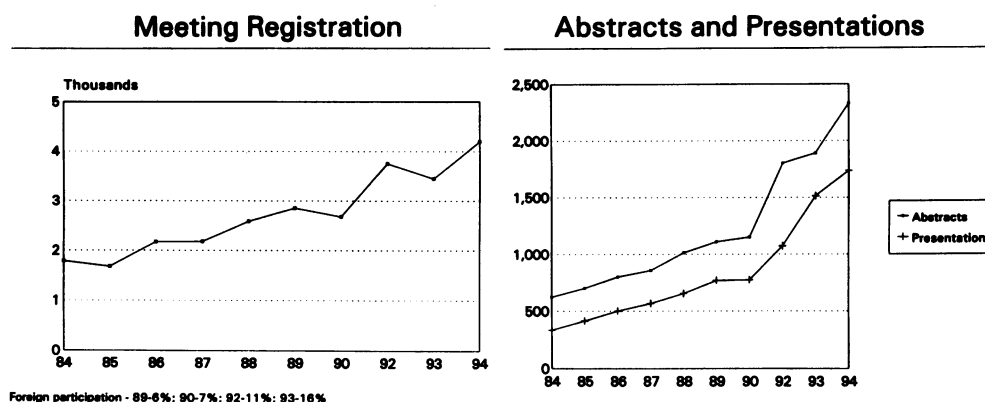
nature) and an increase in applied laboratory research. This raises a concern, since these increases come at the expense of the number of members involved in basic laboratory research. The distribution of laboratory effort demonstrates that a similar number of members are involved in cytogenetics (37%) and molecular genetic (32%) laboratories, while the same percentage of the membership (11%) is working in either biochemical genetics or AFP/triple-screen analysis. Five percent of the membership is involved in newborn-screening laboratories. Comparison of the salary levels across the three surveys also confirms the maturing of the Society, with a "shift to the right" illustrating higher compensation.

The Society's mission is to provide leadership in research, education, and service in human genetics, by providing an interactive forum for sharing research findings affecting individuals and families with inherited conditions. This is accomplished primarily through three modalities:

- *The American Journal of Human Genetics*,
- the annual meeting,
- the activities of the standing and ad hoc committees.

*The American Journal of Human Genetics*, now in its 45th year, is the premier journal publishing research findings, Society position statements, editorials, and announcements. Among the many new genetics journals of both general and specific interest, the "black journal" has maintained its supremacy, reflecting the high standards of the editors and editorial boards.

Individual subscription rates are increasing annually and reflect the Society's growth (fig. 1). The number of pages devoted to scientific papers appears to have gone down, but this may reflect changes in the *Journal's* format. As a matter of fact, since July, each page (even in the enlarged format) holds 12% more material. The real point of figure



**Figure 2** Annual meeting participation. *Left*, Total number of registrants. *Right*, Number of abstracts submitted and presentations of all types.

1 is the number of institutional subscriptions. In the present environment of financial constraints on libraries, to maintain such a distribution record is a testimony to the *Journal's* quality and worth. I might interject at this point that, at the Society's founding in 1948, annual dues for a regular member were \$8.00, including a subscription to the *Journal*. Today's dues of \$90.00, of which \$35 per membership goes toward *Journal* expenses, is still one of the best bargains to be found. In view of inflation during this period, I doubt that the real cost has changed significantly in the past 45 years. Details of this year's *Journal* operation, the first year under its new editor, Peter Byers, will be presented at the membership meeting on Friday.

The annual meeting is the highlight of the administrative year. Its success is easily demonstrated by attendance (fig. 2, left) over the last 10 years, which represents a 135% increase: 1991 has been omitted from this figure, since it was the year of the VIIIth International Congress and not the Society meeting. Increased foreign participation at the annual meeting in the past 4 years is also apparent—from 6% to 16% of attendees.

The numbers of abstracts and presentations have increased accordingly (fig. 2, right). A 237% increase in abstracts received over this same 10-year period is obvious, while the acceptance rate of all types of presentations remained relatively constant at approximately 70%. It is also of interest that 33% of the abstracts this year were foreign submissions.

The duration and program of the meeting are crafted in response to members' requests for innovative formats, such as the thematic public-awareness seminars initiated in New Orleans and continued here in Montreal. Similarly, the Information and Education Committee's project to acquaint high school students with human genetics, possibly as a career choice, has been so successful for the past 2 years that one hopes it will become a standard component of the program. This year there is a half-day Presidential Media Seminar for science writers, on the topic of genetic testing. Incidentally, I highly recommend to all of you

Arno Motulsky's invited editorial on this topic, in the current issue of the *Journal* (Motulsky 1994).

The annual meeting serves yet another vitally important function: one only has to glance at the week's program to appreciate the scope of activities that occur in conjunction with the scientific program. There are 115 other meetings taking place (table 3), including those of the boards of directors of the Society, the American College of Medical Genetics (ACMG), the certification boards, and COMGO, in addition to the active committees and subcommittees of these groups. The hectic work pace of this week is somewhat balanced and moderated by the reunions, receptions, and social gatherings of the various training programs, which give this meeting a unique character.

All of this notwithstanding, the significance of the annual meeting is, of course, its excellent scientific program and the recognition of members' research contributions through individual presentations, the Allan Award Address, awards to our trainees at all levels, and the Distinguished Speakers Symposium. The success of the meeting, year in and year out, is the product of the tireless activities of the Program Committee, which labored for over a year to produce the special sessions, symposia, and workshops that we all enjoy and which had the arduous task this year of sorting through the 2,330 abstracts for presentation. To deal with the ever-increasing number of abstracts, this committee will be enlarged by 33% and already has begun to plan next year's program.

A major contribution to the success of the meeting, and one that is often overlooked, is the untold number of hours invested in the logistics, which assure the smooth functioning of such an event. Here I—we all—must recognize Marsha Ryan and the Society administrative office staff and Nan Nootenboom of FASEB, who actually began planning this meeting in 1988, when Montreal was chosen as the site.

#### Society Activities

It is, perhaps, in the delivery of genetic services and their spin-offs that the Society has undergone the greatest ex-

**Table 3****Ancillary and Additional Meetings Occurring at 1994 Annual ASHG Meeting**

	PROFESSIONAL ORGANIZATION								
	ASHG	ESHG	ACMG	ABMG	NSGC	ABGC	COMGO	CORN	APA
Board of directors .....	+	+	+	+	—	—	+		
Business .....	+		+	+	—	—			
No. of committees .....	10	1	18	3	1	1		2	2
Editorial Boards	Ancillary Meetings/Workshops—29								
American Journal of Human Genetics	Single Disease Groups—14								
Human Molecular Genetics									
Journal of Medical Genetics	Training Program Reunions/Socials—22								
OMIM									

NOTE.—Editorial board meetings were held by *The American Journal of Human Genetics*, *Human Molecular Genetics*, the *Journal of Molecular Genetics*, and *On-Line Mendelian Inheritance in Man*. There also were 29 other meetings and workshops, 14 single-disease group meetings, and 22 training-program reunions/socials.

pansion in the last decade. I remember very clearly, when I assumed the office of treasurer in 1982, the financial records of the Society were kept manually in a notebook given to me by Lillian Lockhart. This binder has since been enshrined in a place of honor in the Bethesda office. At that time, there was a mandate to move the Society to a central office rather than having it administered from an individual member's academic office. The move to the FASEB campus, with an appropriate administrative structure and sufficient staff and space, facilitated the rapid expansion in activities that has occurred. I would like to review some of these major changes contributing to the Society's current situation.

In truth, the process actually began in 1979, with the founding of the National Society of Genetic Counselors (NSGC). Its creation was based on the advances in basic genetics of the 60s and 70s and the need to apply this information to clinical activities. This need could not be met by the existing geneticists, giving rise to a new genetics-health-care professional—the genetic associate or counselor.

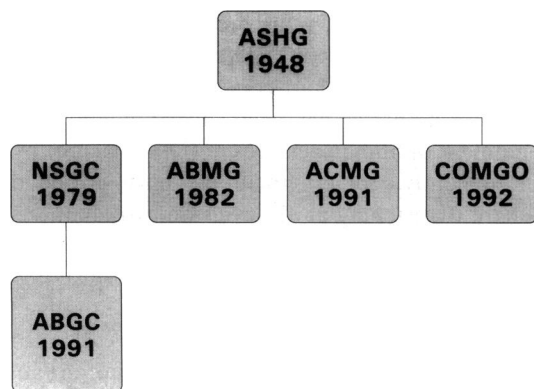
Soon thereafter, in 1982, official recognition of medical genetics training was achieved through the establishment of the American Board of Medical Genetics (ABMG) to accredit training programs and certify individuals. The ABMG provided a framework for and created the professional standards to be met by those wishing certification to practice in the field.

It was also during this period that an organized lay advocacy constituency was established, initially represented by many single-disease parent-support organizations, which subsequently united in the Alliance of Genetic Support Groups, established in 1986 and now comprising more than 200 member organizations. At the same time, the public-health component of the genetics community was being organized, initially on an individual regional ba-

sis, but, beginning in 1985, through the Council of Regional Genetics Networks (CORN). CORN has been supported, since its inception, by the Maternal and Child Health Branch of the Department of Health and Human Services. In this historical overview, we cannot overlook the importance of the Maternal and Child Health Branch, which has been responsible for human genetics training programs at least since the early 1960s. In addition, the SPRANS program (Special Projects of Regional and National Significance) of the Maternal and Child Health Branch has long been a source of funding for clinical research and training programs in human genetics.

Aside from its longstanding record of support for basic and clinical research, perhaps the greatest recent governmental influence on human genetics, through the NIH, has been the establishment of the Human Genome Project as an extramural effort in 1990, with its intramural component initiated last year. This, more than any other single event, provided a significant increase in support for human genomic research. At the same time, it opened the doors for basic scientists outside the established discipline of human genetics to enter the field. The uniqueness of the Human Genome Project's ELSI component, with its direct Congressional mandate to address the nonbiological implications of human genetic research, has placed human genetics, in its broadest sense, in a central position in the nation's scientific efforts.

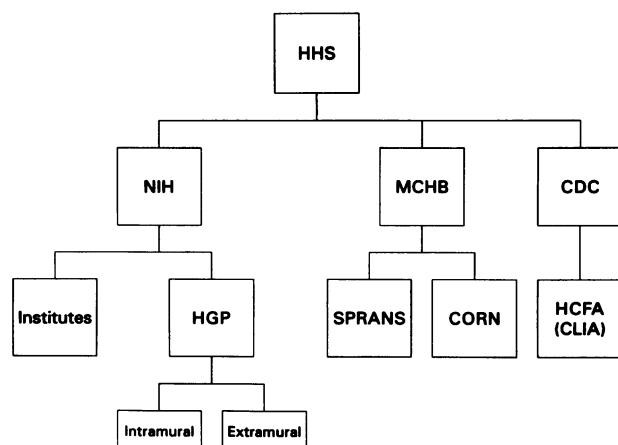
Two final events, both occurring in 1991, complete the development of the current professional structure of the discipline of human genetics. These were the founding of the ACMG and the admission of the ABMG to the American Board of Medical Specialties, the latter recognizing medical genetics as a primary medical specialty and giving equal footing to both M.D. and Ph.D. practitioners. As a result of this last occurrence, the newest member of the professional genetics community was established—the American Board of Genetic Counseling (ABGC).



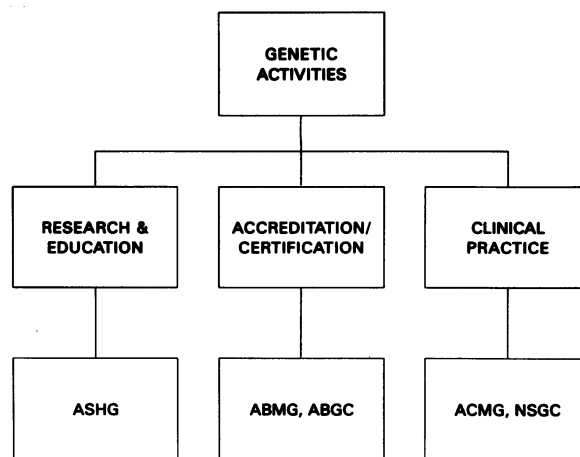
**Figure 3** Pedigree of the professional organizations of the genetics community.

The Society has had significant input into all of these events, through direct monetary support, the provision of the resources and facilities of its office and staff, and, most important, the contribution of individual Society members who provided the leadership for this expansion. As a result, today we operate in a vastly expanded, highly complex, but hopefully integrated genetics community. The pedigree of included professional organizations is represented in figure 3, which has been constructed to show chronological order of creation.

Some of those governmental organizations in the Department of Health and Human Services having the most direct impact on the activities of the Society's members are described in figure 4. Not depicted but obviously important additions are other regulatory components of federal (FDA) and state government and representatives of the executive branch—the Office of Technology Assessment, the Agency for Health Policy Research, the Office of Science and Technology Policy, and professional groups such



**Figure 4** Governmental agencies in the Department of Health and Human services most closely involved with human genetics activities.



**Figure 5** Organizational responsibility for the various human genetic activities.

as the College of American Pathologists and the Joint Commission on Accreditation of Hospital Organizations. In addition, there are a multitude of private foundations, providing research funding and advocacy groups, with whom the Society must interact.

These events have now provided the field of human genetics with a professional organizational structure (fig. 5) similar to that of other medical specialties, with appropriate organizations for oversight of research and educational efforts (the Society), accreditation and certification of training (the ABMG and the ABGC), and clinical practice (ACMG and NSGC). Until quite recently the Society, as the oldest and most established of the groups, had been filling most of these roles alone. This new professional structure and the activities of those interested government and lay groups have made it necessary for the Society to rethink its mode of operation and scope of activities.

From its present level of organizational maturity, the Society must now chart a course toward the 21st century, which will not be subject to the predominantly internal factors that influenced our growth in the past 15 years, but to external pressures that are already playing a more influential role.

As Victor McKusick has repeatedly stated, "All of medicine is genetics." We are perhaps witnessing the extension of that concept to "all of biology is genetics," by which I mean that the study and practice of human genetics can no longer be considered the sole purview of the professional human geneticist. Our specialty enjoys the distinction of converting basic research advances to clinical practice, with a speed unparalleled in any other field of biomedical science. This raises problems, opportunities, and serious challenges, which must be met if the Society is to continue to fulfill its responsibility to both the profession and the various constituencies it serves. The expertise and talents of the genetics community are extremely broad and exten-

**Table 4****Strategic Planning Task Force Committees and Chairs**

Committee	Chair
Basic Research .....	Susan Naylor
Clinical Research .....	Charles Epstein
Education .....	Joann Boughman
Foreign Affairs .....	Walter Nance
Political Action .....	Kurt Hirschhorn
Social, Ethical Issues .....	Phil Reilly

sive. However, a critical caveat guiding our future activities must be the avoidance of redundancy of effort, which would only diminish our impact. This calls for an active program of cooperation and collaboration among *all* the groups involved. In view of the close relationship among many of these organizations, this will be an evolutionary process, demanding time and effort from each group to chart its own course and implement the necessary working liaisons with the others.

The Society has, in point of fact, already instituted such a process during the past 18 months. As president-elect, I was charged by Dr. Janet Rowley to initiate a strategic planning effort whose goal was to evaluate the redistribution of effort necessary to make for more efficient interaction with all members of the genetics community. At the board of directors meeting in the Spring of 1993, a task force of committees (table 4) was established to deal with topics such as basic and clinical research; education and training; social, ethical, and public-policy issues; and foreign affairs. The deliberations of these committees during the remainder of that year were presented for detailed discussion at a 2-day retreat, in February 1994, and for further discussion by the board of directors, in March 1994.

As a result, several suggestions for redirection of Society activities were forthcoming. Perhaps the most significant outcome is the need for the Society to take a more proactive stand on those issues directly affecting its future. This was amply demonstrated during this past year, in those activities in which the Society had traditionally, more or less passively, participated—such as signing on to support increases in the Congressional budget allocations for research and education. This year, however, was extremely exciting because of the possible direct implications of health care–reform legislation and the suggested “pause” on indirect costs, both efforts that were ultimately defeated. Such “legislative” issues, and many others that have an impact on genetics, clearly demonstrate the need for a more politically active stance, as well as the need for the Society to assume a leadership role in keeping the media informed on relevant genetic issues. Additionally, a realignment of the yearly cycle of the Society’s committees was designed and implemented to provide greater effi-

ciency in drafting policy statements to allow for more intensive—and significantly shorter—periods of activity.

It is perhaps in the area of education that our emphasis and scope of activities has been most influenced by the new alignment of human genetics professional groups. Until quite recently, Society members have provided the lion’s share of teaching at all levels and to all audiences. This situation has changed rapidly with the development of a multifocal genetics community, each speaking to a more directed constituency. These audiences range from primary school through professional trainees seeking medical genetics board certification, to parent-support and advocacy groups and the general public.

A new area of activity suggested by the long-range planning program was to become more involved internationally in human genetics. With the significant increases in foreign membership and overseas participation at the annual meeting, we should perhaps initiate an effort for more international interaction and organization.

### **How Has the Society Responded to These Suggestions?**

The following actions have resulted from a series of meetings with representatives of the other organizations and, most important, deliberations among the Board of Directors and its Executive Committee:

#### **Public Policy and Political Action**

The intensive efforts by the many groups attempting to shape health care–reform legislation clearly demonstrated the need for a proactive role for the Society—as well as the entire genetics community—if our concerns are to be addressed. The current Society machinery is far too slow to provide the rapid responses necessary for many of the actions that must be taken. The Public Policy Committee, therefore, has been restructured to function as a fast-track response team for issues arising between meetings of the Board of Directors meetings. The committee’s new membership consists of the past president as chair, the officers, chairs of the Social Issues and Human Genome Committees, two board members, and a member-at-large. Ad hoc consultants will be added as necessary for the specific issues.

It is also clear that we are relatively uninitiated in political activity, which will demand an ever-increasing effort on our part. To this end, the Board of Directors has approved the establishment of an AAAS Congressional Fellowship in Human Genetics. This fellowship, when implemented, will be jointly sponsored by the Society and the Human Genome Project of the U.S. Department of Energy and will allow genetics professionals to spend a year as a legislative assistant to a member of Congress or serve on the staff of a Congressional committee. It is designed to educate human-genetics health-care professionals in the legislative

process and the field of policy development. Hopefully, in time, graduates of this program will provide the Society with a cadre of knowledgeable, experienced members who can serve as a ready resource in the development of public-policy positions. Application procedures for this fellowship, which will be instituted in the fall of 1995, will be distributed to the membership shortly.

Of a more immediate and practical nature is the need for an in situ professional geneticist to represent the Society's viewpoint at the multitude of important meetings occurring in Washington, D.C. throughout the year. While, at first glance, many of these meetings may not appear to directly involve genetics, they actually impinge significantly on our research and educational missions. In addition, such an individual will work closely with various committees in drafting position statements and facilitate their timely development and completion. For example, this year position papers developed by Society committees dealing with (1) insurance issues and (2) screening for breast/ovarian cancer have been completed and submitted to the *Journal* for publication. Closer oversight of such activity may have led to a more timely appearance of these statements. This, after all, is an important component of the Society's activity. I am pleased to announce that the Board of Directors has approved the hiring of a part-time public-policy consultant in the administrative office, to fill this role.

Another critical aspect of the public-policy arena is the monitoring of state and federal legislation on genetic issues. We have attempted to assess the scope of the problem by taking advantage of a computerized legislative monitoring service at BIO (the Biotechnology Industry Organization). Screening for only two issues was extremely enlightening. Using the key words "DNA testing/forensics/identification," 36 proposed bills in 16 states were found; under "genetic screening/discrimination" there were 20 proposed pieces of legislation in 18 states. To keep on top of these issues, the Society will make arrangements with an established legislative monitoring service for genetic issues, to provide us with this information on a regular basis. An important but as yet unresolved issue is how to mount a meaningful response once such information is in hand. Preliminary discussions with representatives of CORN and COMGO have begun to design grassroots mechanisms for making our collective voice heard.

A final item in public-policy activity includes two articles dealing with genetic testing that are currently being developed by members of the Board of Directors. One article is to be directed at the "Perspectives" section of *Science*, for the professional audience, and the other is an op-ed piece for the lay public.

#### **Other Committee Restructuring**

I have already mentioned the new look of the Public Policy Committee. In addition, the Board of Directors has

decided that one of its members should serve on each Society committee, to provide closer liaison between the committees and the Board.

The following Society committees will retain their present composition and continue to function as presently described in the bylaws: Awards, Executive, Finance, Nominating, and Program. However, changes have been suggested for some committees, and several new ones have been formed.

#### **Social Issues Committee**

Inasmuch as many of our sister organizations have similar committees, it was suggested that the Social Issues Committees of the Society, the ACMG, the NSGC, and the International Society of Nurses in Genetics (ISONG) meet to coordinate agendas, being particularly sensitive to the possibility of joint projects and eliminating overlap of effort. The first meeting of this combined group took place on Tuesday of this week, and it is hoped that such a meeting will occur annually. Currently, there is a "points to consider" project on genetic testing in children, in preparation by this committee.

#### **Genome Committee**

While maintaining its present activities and serving as the Society's liaison to the Human Genome Project via its chair, the genome committee will assume an additional role. The long-range-planning task force on research has strongly suggested that defining and/or directing specific areas of research to be pursued is not the function of the Society. However, it is indeed appropriate to monitor research developments that will have a direct impact on Society activities. The two research areas suggested for close monitoring by the genome committee are reproductive technologies and gene therapy, with the hope that appropriate and timely positions and statements on these issues, representing the Society's stand, can be developed.

#### **Information and Education Committee**

With the present lineup of organizations involved in education, efficiency dictates a certain degree of specialization. In response to the long-range-planning committee, the Board of Directors, and a Joint Executive Committee of the Society and the ACMG, the following scheme has been suggested. The Society should take the lead in K-12, undergraduate, and graduate education, including predoctoral M.D. and postdoctoral research training. The ACMG and the NSGC would be primarily responsible for those training programs involving professional ABMG and ABGC certification.

With regard to nongeneticist education, the Society should take the lead in trying to introduce basic genetics into the curriculum of nurses. Such a dialogue has already been initiated with Cynthia Prows, President of ISONG, who sent a representative to the Information and Educa-

tion Committee meeting. Educational efforts for the lay public are the responsibility of all but should be issue-specific, with shared leadership where appropriate.

**Executive Committee.**—*The Executive Committee Will Hold Joint Meetings with its Counterparts in the ACMG and the European Society of Human Genetics, on an Annual Basis.*

#### New Committees

The establishment of several additional Society committees has been recommended:

**Test and technology-transfer committee.**—At the Joint Executive Committee meeting of the ACMG and the Society, held in July, it was recommended that a joint advisory committee be established to address focused issues involving the transfer of new technologies and tests into clinical service. This committee will be cochaired by a Society member and an ACMG member selected by their respective Boards of Directors and will comprise equal representation from both organizations. It would establish working groups to address specific tests or issues. Mechanisms are currently being developed with regard to which organization should have final responsibility for position papers and/or policy statements on genetic testing. Liaison and input from other interested organizations are expected at all levels of review.

**Foreign affairs committee.**—In keeping with the recognition of increased foreign membership, an effort should be made to intensify interaction with other genetics societies around the world. This past year, a delegation of 30 geneticists, mostly Society members, went to China and Mongolia on a people-to-people mission under the leadership of our past president, Walter Nance. This visit opened new lines of personal communication and led to the sending of a significant number of recent genetics journals to the library of the medical school in Ulan Bataar, Mongolia.

At last year's annual Society meeting, Dr. Albert de la Chappelle, then president of the European Society of Human Genetics, attended our Board of Directors meeting. This past June, I was invited to give a presentation to the European Society of Human Genetics annual meeting on the organization of the U.S. genetics community. On that occasion, I also participated in their board of directors meeting. This past Monday, there was a meeting of the combined Society/ESHG executive committees, which investigated ways of further strengthening ties between these two societies and, possibly, others throughout the world.

**Database committee.**—A recent national meeting, organized under the auspices of the Biophysical Society, at which we were represented by several members, raised problems relating to the development and maintenance of molecular biological data bases. Critical issues concerning

the scientific community's education and awareness of these data bases, their maintenance, confidentiality, and access, were discussed at length. From the Society perspective, there are a variety of additional data bases that would be useful to our members. These are not only research tools, but contain much useful and important information pertaining to the clinical and educational realms as well. Therefore, the Society has established a broad-based ad hoc committee charged to study human genetic data bases. In collaboration with the Information and Education Committee, the data base committee will also address issues associated with informing the membership of these important data bases and educating us as to their use.

**Bylaws committee.**—Finally, to deal with the formal establishment of the new committees and the restructuring of existing ones, as well as to accommodate several other governance changes currently being discussed by the Board of Directors, a bylaws committee will be established. This committee will examine and suggest necessary changes to accommodate the new structures, hopefully leading to ratification by the membership and to more efficient operation.

These realignments will allow for appropriate responses to issues we will face in the *immediate* future. I would like to spend the next few minutes addressing those areas that I consider will have a longer-term impact on the Society and that demand attention and planning now, so that we will be positioned to act rather than react. All of these issues will impact significantly on research and education, the principal *raison d'être* of the Society.

The first issue is the present and future environment for research in human genetics. There is no need to discuss the budgetary constraints being placed on publicly funded research. The increasing numbers of well-qualified investigators competing for a shrinking pool of resources has already influenced and will increasingly influence individuals contemplating careers in academic research. This situation has been exacerbated by the unparalleled excitement generated by advances in genetics that have attracted many highly competent researchers, who themselves are not trained as human geneticists but who nonetheless increase the level of competition.

A second influence, and perhaps one of greater concern, is the amount of basic human genetic research being conducted in the private sector. Estimates indicate that the research and development budgets of the 15 largest pharmaceutical companies is approximately \$12 billion dollars—roughly equaling the *total* NIH budget. Similarly, the aggregate research and development budgets of all biotech companies combined ( $\sim 1,300$ ) is \$5.7 billion, with the largest 150 contributing \$3.1 billion of this sum. It is also estimated that more than 50% of *basic* human genetic research is being carried out by commercial concerns. This is *not* directed, product-oriented application, but extremely sophisticated top-quality basic science. The open-

ness and sharing of information so common in public science is incompatible with the corporate philosophy. Furthermore, the proprietary nature with which investigative results are currently being considered by a growing segment of the research community is indeed a general concern. The recent stand by Dr. Varmus and the NIH, regarding the patenting of cDNA sequences and Merck's recent grant to Washington University to sequence 200,000 cDNAs in the next 18 months, for immediate contribution to GenBank, are to be applauded. However, an increasing number of our members are filing for individually held patents, encouraged by the technology-transfer offices of their universities. More members are now full- or part-time employees of private companies, to say nothing of the large and growing number of corporate consultants among the membership. These topics have been discussed in the commentary sections of recent issues of *Science and Nature* and make most interesting reading.

The obvious effect of these developments may lead to conflict of interest or its appearance, resulting in a multiplicity of disclosure statements (possibly even on NIH grant applications), proprietary restrictions on information and reagent exchange, and ultimately to increased cost of research, because of patent and licensing fees. These two factors are not unrelated: with diminished support of public research funding directly translating to greater competition, some of our most dynamic and original researchers are being attracted to the fast-paced and lucrative biotechnology industry.

Activity of the private sector also has a profound effect on training in human genetics. Not only are the emerging companies attracting our trainees to perform research, but their direct competition for provision of genetic services and testing threaten the financial underpinning of training programs. Companies that provide both laboratory and clinical services are successfully competing with academic institutions that have traditionally relied on clinical income to fund fellowships and stipends for trainees. In essence, this results in the doubled effect of both absorbing the graduating trainees and negatively affecting the means for their support during training.

These issues of a commercial nature introduce another related concern, which will change the way medicine is practiced—and human genetics along with it. I am referring to the expansion of managed-health-care schemes and their influence on academic medical centers, where the majority of the Society membership is still employed. The most direct and obvious effect is the reduction of clinical practice income in the academic centers, which are non-competitive in service delivery, because of the added expenses of research and teaching. The loss of these resources is already being felt in every department in every medical school across the country. The need to change the traditional fee-for-service approach requires a paradigm shift for specialty services, if they are to survive in this new

environment. There are no obvious or easy solutions for this dilemma, since in many cases it is institution specific. However, it clearly behooves the human genetics community to create, mold, and implement the appropriate academic/corporate collaborations that might define a win-win situation.

Coupled with direct financial loss is the thrust to limit specialist training and the call to produce an increased number of generalist physicians. For us, as the newest of the primary medical specialties, this directive could not have come at a more inauspicious time. Before programs can even become established under the new format, specialty training is being deemphasized. This poses yet another related problem—that is, the genetics education of those generalist M.D.'s. Not only is the frequency of gene discovery increasing, but we are also beginning to identify those genes whose prevalence in the population will assume significant proportions. The recent discoveries of the genes for various familial neoplasias may already have initiated this new era. As the genes responsible for the more common diseases are elucidated, genetics will assume a much more prominent position in the public-health arena. Since the primary-care providers will serve as the health-care gatekeepers for an ever-increasing number of patients demonstrating genetic diseases, providing them with the knowledge necessary to successfully interact with their patients will be a monumental task.

The third and perhaps the most perplexing and complicated of the external influences we face in the next decade will be the legislation of genetics. The failure of health-care reform perhaps provides a portent of things to come. This was undoubtedly the most heavily lobbied piece of legislation in U.S. history, its price tag mounting into the hundreds of millions of dollars. The failure of Congressional action was directly attributable to the success of a multitude of diverse special interest groups, each attacking a different aspect of each bill. The outcome could have been predicted. Needless to say, this victory of the vested-interest groups provides a successful game plan for interested parties to follow in the future. Efforts in this direction are already apparent in legislative programs at all levels of government. A few examples among the many that will have to be faced are the mandated creation of DNA repositories, the blurred delineation of clearly service laboratories from those providing patient results on an investigative basis, and the need for licensing such labs, not to mention those familiar issues that fall under the rubric of the ethical, legal, and social implications of genetics research—confidentiality, informed consent, discrimination in the workplace, and health insurance. There are no easy or ready-made strategies to withstand the external forces that will certainly shape the Society's actions, and perhaps even its character, in the coming decade. Nor is there any time to lose in fashioning approaches to deal with them. Hopefully, an active continuation of the plan-

ning process, with broad membership participation, will provide the necessary framework to do so.

I hope that it is very clear that the environment in which we currently operate has significantly changed in the past decade. Moreover, this process of change will continue for the foreseeable future. Some of the factors that will effect these changes are obvious, and I have tried to enumerate three of the most acute. Undoubtedly, there will be others, of which we are currently unaware, that will also play a role. However, I see this situation as not posing insurmountable problems, but, rather, as creating new challenges to be confronted with the same vitality and inventiveness that allowed the most impressive Society achievements of the past 10 years. This, however, will demand a closer collaboration among all members of the genetics community—and mutual helpfulness.

In closing, I would like to say that it has truly been a

privilege to serve as your president this year. Not only was it an education, but along the way I got to know a lot of you much better. This Society has a great wealth of expertise residing in a large number of very talented members. Although the work was sometimes pressured and crisis-laden, my colleagues on the Board of Directors and the active members of the committees were always there to help meet the challenges. More than that, the excellent staff of the administrative office, most ably led by Elaine Strass, made the work a pleasure. I thank you.

## References

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